

GENE NEWS

A Publication of the Hawai'i Department of Health Genetics Program



FALL 2004 CONFERENCE

Friday, September 10th, 2004

7:30am - 3:30pm

Pacific Beach Hotel - Waikiki

The Hawai'i Department of Health Genetics Program is delighted to announce this exciting one-day conference targeted towards allied health professionals, public health professionals, physicians in all specialties, and students in these disciplines. Speakers consist of physicians from the mainland, as well as local physicians and genetics professionals. The conference will:

- Increase appreciation for genetic disorders or disorders with a genetics component.
- Provide knowledge and tools to identify and make appropriate referrals to genetics.
- Increase knowledge of genetics related projects in Hawai'i and local resources.

The cost of registration is \$25 for allied health professionals, and \$50 for physicians. The registration deadline is August 31st, 2004. Please contact Sharon (sharon@hawaiiigenetics.org or 733-9055) to request a conference brochure, or Allison (allison@hawaiiigenetics.org or 733-4998) with any questions. We look forward to seeing you on September 10th!



**GENETICS
FOR YOUR
PRACTICE
INFORMATION
AND TOOLS
FOR ALLIED
HEALTH
PROFESSIONALS
&
PHYSICIANS**

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Coordinator's Corner

I want to encourage you to attend the Genetics Conference on September 10th. We are listening to your comments from the 2003 conference and making this conference as useful to YOUR daily practice as possible. We want to make sure you have the cutting edge genetics information and tools to use for your clients and patients. We are only having the conference on one day this year so make sure to register early to guarantee your space.

We are pleased to announce a new section for teachers that will be included in every issue of GeneNews. This section will highlight information that is useful for those who teach genetics at every level.

Please let me know if you have any ideas for information you want to see in future issues or have comments about Gene News in general.

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*In May 2004, the Minneapolis Star Tribune newspaper ran a story about a family struggling with a physician's **duty to warn** about a genetic disorder.*

According to the article, Mrs. X had a three-year old daughter, Jane X, who was developmentally delayed. The pediatrician mentioned a genetic cause as a possibility, especially since Mrs. X's half-brother has mental retardation. The pediatrician then told Mrs. X that she would run a series of tests on the daughter, including the genetic test for Fragile X syndrome.

People with Fragile X syndrome generally have some degree of mental retardation. All mothers of a child with a full gene mutation are carriers, meaning that they can pass the mutation on to their children. The mother and her family members are at increased risk of having children with Fragile X syndrome, and should be offered genetic counseling and testing.

The test results for Jane X came back, and were reported to Mrs. X as normal; Mrs. X assumed that this included the test for Fragile X. She was not aware, however, that the Fragile X test results were not included in the lab results. Mrs. X went on to have three more children, none of whom had developmental delay. Her fifth child, a son, was born with Fragile X syndrome, and only then did Mrs. X learn that she is a carrier of the gene mutation.

Mrs. X reported that she and her husband would have chosen sterilization if she had known she was a carrier for the gene mutation. She sued. However, the doctor claimed that her responsibility was only to her patient, Jane X, and not to Jane's mother, Mrs. X. Based on this assumption, the doctor wanted the case dismissed.

However, the Minnesota Supreme Court agreed with the lower courts and would not dismiss the case. The court stated that "...a physician's duty regarding genetic testing and diagnosis extends beyond the patient to the biological parents who foreseeably may be harmed by a breach of that duty. In this case, the patient suffered from a serious disorder that had a high probability of being genetically transmitted and for which a reliable and accepted test was widely available. The appellants should have foreseen that parents of childbearing years might conceive another child in the absence of knowledge of the genetic disorder." Mrs. X is free to continue with her lawsuit.

One of the lawyers for the physician called the decision "a troubling precedent", and said that it could lead to lawsuits from relatives claiming harm because of a missed diagnosis. "Many genetic conditions or diseases skip a generation or don't appear until the next generation....That doctor will remain liable forever."

This case is an example of how complex the world of medical genetics often is. Do you think Jane X's pediatrician should be held liable? Do you think that Mrs. X would have chosen a different reproductive future if she had known she was a carrier? Do you think that a doctor should share his patient's genetic test results with that patient's family members?

There are many ethical, legal, social, and emotional issues to be considered as more genetic tests become widely available. Ideally, genetic test results should be discussed with a genetics professional, or a health professional with training in genetics, and referrals to genetics services should be made when appropriate.

The Pandora's Box of Genetic Testing "Duty to Warn"

*Test Results
Duty to Warn
Reproductive Choice
Duty to Recontact
Genetic Testing*





Sometimes Less is More... Newborn Metabolic Screening Brochures



If you are an expectant mom or dad trying to learn about Newborn Metabolic Screening (NBMS), you will no doubt receive a blue brochure full of details and descriptions. Although the brochure contains several adorable pictures of babies, it likely won't take long for the text-heavy panels to deter you from your reading—especially if you are among the 18% of adults in Hawai'i with low literacy.

When the Hawai'i Genetics Program asked moms and dads to tell us what they thought about this NBMS brochure, we received responses like, "The brochure is boring," "It has too many words," and "I would never read this."

In an attempt to make the important newborn screening information more appealing to parents, the Genetics and NBMS Programs have developed a shorter, more appealing NBMS brochure based on suggestions made by moms and dads in focus groups and on surveys. Unlike the monochromatic blue of the current brochure, the new brochure is full of pastel colors, is only one panel long, and is much simpler in format.

Our next question was, "Do these shorter, straightforward brochures still contain enough information to educate moms and dads about NBMS?" To answer this, we took our brochures to three Women, Infant and Children (WIC) offices on O'ahu. At the WIC offices, we asked a total of 102 parents to fill out a survey upon reading the new brochure. We then asked an additional 100 WIC parents to read the current blue brochure and fill out the same survey.

Survey analysis revealed that there was no statistically significant difference in the amount of information contained in the two brochures. In addition, approximately 14% of parents who read the current brochure said that it included too much information compared with only 2% of parents who read the new brochure. Finally, the majority of parents (96%) believed that the new brochure was easy to read, and 84% said they would pick up the new brochure while at their doctors' offices.

We are excited about the positive responses regarding our new NBMS brochure. The new brochure is an additional resource that will be distributed to clinics, offices, and other community health facilities in the near future. Thank you to all of the WIC offices and parents who helped us with our surveys!



Teacher's Corner

Welcome to a new continuing column in GeneNews. We have received much positive feedback from teachers who use Gene News as a resource.

Several teachers have suggested that Gene News would be a great place to include news specifically for teachers: new developments in the world of genetics, good educational resources, educational websites, and upcoming conferences or workshops. We plan to include these items in future issues and hope that teachers will continue to utilize Gene News as a resource when planning genetics lessons. If you have any contributions you feel would be appropriate for upcoming Teacher's Corner sections, please contact Kirsty McWalter at kirsty@hawaiigenetics.org.

Clinic Schedule – Fall 2004:

- **August 9th - 13th, 2004**
Dr. Greg Enns
Metabolic Clinic
- **September 13th - 17th, 2004**
Dr. Gene Hoyme
General Genetics Clinic
- **October 18th - 22nd, 2004**
Dr. Louanne Hudgins
General Genetics Clinic
- **November 15th - 17th, 2004**
Dr. Melanie Manning
General Genetics Clinic
- **December 13th - 17th, 2004**
Dr. Gene Hoyme
General Genetics Clinic

Hawai'i Community Genetics is a collaborative program involving *Kapi`olani Medical Center for Women & Children, Queen's Medical Center, the University of Hawai'i John A. Burns School of Medicine, the Department of Health, and Hawai'i Medical Services Association*. Geneticists from *the Stanford School of Medicine* visit Hawai'i approximately one week per month for pediatric and adult genetics clinics, as well as to provide genetics education to various groups. Since the clinics began in June 2003, Hawai'i Community Genetics clinics have been filled with patients, and referrals to the program continues to increase. The box to the left is a listing of upcoming clinics. Referrals to Hawai'i Community Genetics can be directed to (808) 973-3403.